

Title (en)

FUNCTIONAL GENETIC TESTS OF DNA MISMATCH REPAIR

Title (de)

FUNKTIONALE GENETISCHE TESTS FÜR DNA-FEHLPAARUNGSREPARATUR

Title (fr)

EPREUVES GENETIQUES FONCTIONNELLES DE LA REPARATION DES APPARIEMENTS DE L'ADN

Publication

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Application

EP 02763876 A 20020329

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Abstract (en)

[origin: WO02081624A2] An invention is described which provides a diagnostic approach for diseases, such as HNPCC, that are associated with defects in MMR and provides a method for determining whether any specific genetic sequence of a gene associated with MMR that differs from a consensus sequence is a mutation (i.e. </i>, encodes a non-functional protein), a silent polymorphism (i.e. , encodes a protein with normal protein function) or an efficiency polymorphism (i.e. , encodes a protein with reduced efficiency in MMR). The invention allows the generation of databases of the functional significance of specific amino acid replacements on MMR protein function in vivo , which in turn will allow accurate and unambiguous interpretation of genetic tests of MMR.

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